RELN gene

reelin

Normal Function

The *RELN* gene provides instructions for making a protein called reelin. This protein is produced in the brain both before and after birth. Reelin is released by certain brain cells; then it attaches (binds) to specific receptor proteins. In the developing brain, this binding turns on (activates) a signaling pathway that triggers nerve cells (neurons) to migrate to their proper locations.

After birth, reelin likely plays a role in many brain processes, including the extension of axons and dendrites, which are specialized outgrowths from nerve cells that are essential for the transmission of nerve impulses. Reelin may also regulate synaptic plasticity, which is the ability of connections between neurons (synapses) to change and adapt over time in response to experience. Additionally, reelin controls the release of chemicals that relay signals in the nervous system (neurotransmitters).

Health Conditions Related to Genetic Changes

lissencephaly with cerebellar hypoplasia

At least six mutations in the *RELN* gene have been found to cause lissencephaly with cerebellar hypoplasia (LCH). This condition affects brain development, resulting in the brain having a smooth appearance (lissencephaly) instead of its normal folds and grooves. In addition, the brain region involved in coordinating movements is unusually small and underdeveloped (cerebellar hypoplasia). The *RELN* gene mutations that cause LCH lead to a complete lack of reelin. As a result, the signaling pathway that triggers neuronal migration is not activated. Without reelin, neurons are disorganized, the normal folds and grooves of the brain do not form, and brain structures do not develop properly. This impairment of brain development leads to intellectual disability, delayed overall development, movement problems, and other signs and symptoms of LCH.

other disorders

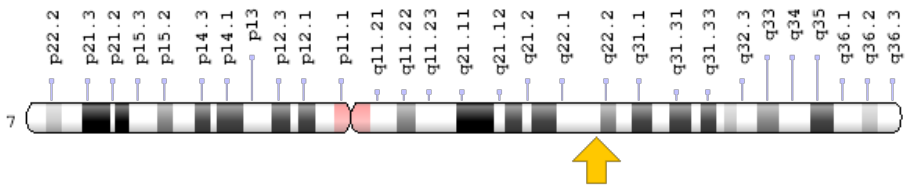
Studies have shown certain variations (polymorphisms) in the *RELN* gene to be associated with an increased risk of psychiatric disorders such as schizophrenia and bipolar disease. Women with these polymorphisms are at particular risk of developing bipolar disease. In addition, certain genetic changes that result in a decrease in production of reelin (but not a complete absence) may be a risk factor for autism spectrum disorders, which affect communication and social interaction. However,

other studies have not supported these findings. Many genetic and environmental factors are believed to contribute to these complex conditions.

Chromosomal Location

Cytogenetic Location: 7q22.1, which is the long (q) arm of chromosome 7 at position 22.1

Molecular Location: base pairs 103,471,784 to 103,989,516 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- LIS2
- PRO1598
- RELN_HUMAN
- RL

Additional Information & Resources

Educational Resources

- Jasper's Basic Mechanisms of the Epilepsies (fourth edition, 2012): Neuronal Migration Signaling Pathways (figure)
  https://www.ncbi.nlm.nih.gov/books/NBK98194/figure/liu.f3/?report=objectonly

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RELN%5BTIAB%5D%29+OR+%28reelin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- REELIN
  http://omim.org/entry/600514

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_RELN.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RELN%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9957
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5649
- UniProt
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